

Brief Clinical Report

Anomalies of the Nipple: An Additional Finding in CHARGE Syndrome

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INTRODUCTION

Pagon et al. [1981] proposed the mnemonic CHARGE for the combination of coloboma, heart disease, choanal atresia, retardation, and genital and ear anomalies. Since then, several additional manifestations have been described in this condition which Lubinsky considers a *syndrome*, not an association. We report on a 5-week-old infant with CHARGE syndrome and another anomaly, hypoplasia of the nipple, which, to the best of our knowledge, has not been listed as an associated finding to date.

CLINICAL REPORT

The infant was born to healthy Ashkenazi Jewish parents after an uneventful 39-week pregnancy. Toward the end of the pregnancy, ultrasonographic examination showed bilateral hydronephrosis and polyhydramnios. Delivery was normal, with a birth weight of 2,990 g, and a single umbilical artery, left facial palsy, asymmetric face, abnormality of the left earlobe, a cleft of the soft palate, unilateral choanal atresia, bilateral coloboma of the choroid, and a systolic murmur grade 3/6, heard best over the pulmonic area. Lungs were clear. Hypoplasia of the left nipple was noted. Genitalia were normal. Echocardiogram demonstrated atrioventricular (AV) canal, patent ductus arteriosus, and pulmonic stenosis with pulmonary hypertension. Ultrasound showed bilateral hydronephrosis, confirmed by a voiding cystourethrogram which showed bilateral vesicoureteral reflux with a right hydroureter. Computerized tomography of the brain showed mild "brain atrophy." Chromosomes were normal (46,XX).

DISCUSSION

Our patient has CHARGE syndrome with hypoplasia of the left nipple. Qazi et al. [1982] described 3 children

with choanal atresia, 1 of whom (a boy) was found to have bilateral athelia, delayed speech development, displacement of the midline structures of the brain, and slightly enlarged lateral and third ventricles. This child may have had a form of CHARGE syndrome. Greenberg [1987] reported on an infant with choanal atresia, delayed motor development apparent at age 6 months, and athelia, and he proposed methimazole teratogenicity as a cause. The infant may also have had CHARGE syndrome.

The present case raises the possibility that anomalies of the nipple are included among the phenotypic anomalies found in correlation with CHARGE syndrome.

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